



## VPS13B gene

vacuolar protein sorting 13 homolog B

### Normal Function

Researchers have not determined the role of the *VPS13B* gene (frequently called the *COH1* gene) in the human body. Studies have shown that it has some similarities to a gene found in yeast that is involved in sorting and transporting proteins inside the cell. The human *VPS13B* gene is normally turned on in most cells of the body, which suggests that it plays an important role in cellular function. It may be particularly important in the formation of blood cells and the development and function of the eye and brain.

### Health Conditions Related to Genetic Changes

#### Cohen syndrome

At least 73 different mutations in the *VPS13B* gene have been shown to cause Cohen syndrome. Most of these mutations result in a premature stop signal in the instructions for making the *VPS13B* protein. Researchers believe that this genetic change leads to the production of an abnormally short, nonfunctional version of the protein. It is not known how the absence of the *VPS13B* protein leads to the signs and symptoms of Cohen syndrome.

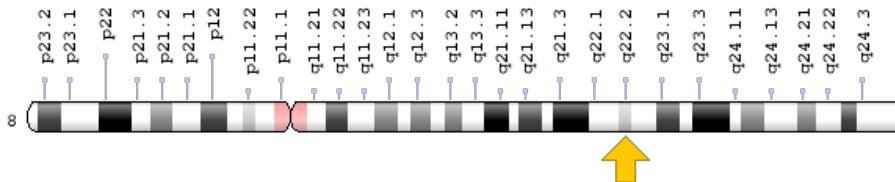
In the Finnish population, 75 percent of individuals with Cohen syndrome have a mutation in the *VPS13B* gene that deletes two DNA building blocks (base pairs). This mutation is sometimes written as 3348\_3349delCT. The deletion causes a premature stop signal in the instructions for making the *VPS13B* protein.

Two common mutations are seen in the Old Order Amish population. The first mutation inserts one base pair and is sometimes written as 9258\_9259insT. This mutation creates a premature stop signal in the instructions for making the *VPS13B* protein. The second mutation changes a single protein building block (amino acid) in the *VPS13B* protein. Specifically, this mutation replaces the amino acid isoleucine with the amino acid threonine at position 2820 (written as Ile2820Thr or I2820T). Outside the Finnish and Amish populations, nearly all mutations in the *VPS13B* gene are seen in only one or a small number of families.

## Chromosomal Location

Cytogenetic Location: 8q22.2, which is the long (q) arm of chromosome 8 at position 22.2

Molecular Location: base pairs 99,007,014 to 99,877,586 on chromosome 8 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- CHS1
- COH1
- Cohen syndrome 1
- DKFZp313I0811
- KIAA0532
- vacuolar protein sorting 13 homolog B (yeast)
- vacuolar protein sorting 13B
- VP13B\_HUMAN

## Additional Information & Resources

### GeneReviews

- Cohen Syndrome  
<https://www.ncbi.nlm.nih.gov/books/NBK1482>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28VPS13B%5BTIAB%5D%29+OR+%28COH1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+NOT+%28%28streptococcus%29+OR+%28streptococcal%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2520+days%22%5Bdp%5D>

## OMIM

- VACUOLAR PROTEIN SORTING 13, YEAST, HOMOLOG OF, B  
<http://omim.org/entry/607817>

## Research Resources

- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=VPS13B%5Bgene%5D>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=2183](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=2183)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/157680>
- UniProt  
<http://www.uniprot.org/uniprot/Q7Z7G8>

## **Sources for This Summary**

- Falk MJ, Feiler HS, Neilson DE, Maxwell K, Lee JV, Segall SK, Robin NH, Wilhelmsen KC, Träskelin AL, Kolehmainen J, Lehesjoki AE, Wiznitzer M, Warman ML. Cohen syndrome in the Ohio Amish. *Am J Med Genet A.* 2004 Jul 1;128A(1):23-8.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/15211651>
- GeneReview: Cohen Syndrome  
<https://www.ncbi.nlm.nih.gov/books/NBK1482>
- Hennies HC, Rauch A, Seifert W, Schumi C, Moser E, Al-Taji E, Tariverdian G, Chrzanowska KH, Krajewska-Walasek M, Rajab A, Giugliani R, Neumann TE, Eckl KM, Karbasiyan M, Reis A, Horn D. Allelic heterogeneity in the COH1 gene explains clinical variability in Cohen syndrome. *Am J Hum Genet.* 2004 Jul;75(1):138-45. Epub 2004 May 20.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/15154116>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1181997/>
- Kolehmainen J, Black GC, Saarinen A, Chandler K, Clayton-Smith J, Träskelin AL, Perveen R, Kivitie-Kallio S, Norio R, Warburg M, Fryns JP, de la Chapelle A, Lehesjoki AE. Cohen syndrome is caused by mutations in a novel gene, COH1, encoding a transmembrane protein with a presumed role in vesicle-mediated sorting and intracellular protein transport. *Am J Hum Genet.* 2003 Jun; 72(6):1359-69. Epub 2003 May 2.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/12730828>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1180298/>
- Kolehmainen J, Wilkinson R, Lehesjoki AE, Chandler K, Kivitie-Kallio S, Clayton-Smith J, Träskelin AL, Waris L, Saarinen A, Khan J, Gross-Tsur V, Traboulsi EI, Warburg M, Fryns JP, Norio R, Black GC, Manson FD. Delineation of Cohen syndrome following a large-scale genotype-phenotype screen. *Am J Hum Genet.* 2004 Jul;75(1):122-7. Epub 2004 May 12.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/15141358>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1181995/>

- Seifert W, Holder-Espinasse M, Spranger S, Hoeltzenbein M, Rossier E, Dollfus H, Lacombe D, Verloes A, Chrzanowska KH, Maegawa GH, Chitayat D, Kotzot D, Huhle D, Meinecke P, Albrecht B, Mathijssen I, Leheup B, Raile K, Hennies HC, Horn D. Mutational spectrum of COH1 and clinical heterogeneity in Cohen syndrome. *J Med Genet.* 2006 May;43(5):e22.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/16648375>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2564527/>
- Velayos-Baeza A, Vettori A, Copley RR, Dobson-Stone C, Monaco AP. Analysis of the human VPS13 gene family. *Genomics.* 2004 Sep;84(3):536-49.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/15498460>

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